

## Product datasheet for **SC314762**

### Ataxin 1 (ATXN1) (NM\_000332) Human Untagged Clone

#### Product data:

Product Type:	Expression Plasmids
Product Name:	Ataxin 1 (ATXN1) (NM_000332) Human Untagged Clone
Tag:	Tag Free
Symbol:	Ataxin 1
Synonyms:	ATX1; D6S504E; SCA1
Mammalian Cell Selection:	None
Vector:	<u><a href="#">pCMV6-XL5</a></u>
E. coli Selection:	Ampicillin (100 ug/mL)



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**Fully Sequenced ORF:** >OriGene ORF sequence for NM\_000332 edited  
GAAACAGTCACCGTGGAGGGGGACGGCGAAAAATGAAATCCAACCAAGAGCGGAGCAAC  
GAATGCCTGCCTCCCAAGAAGCGCGAGATCCCCGCCACCAGCCGGTCTCCGAGGAGAAG  
GCCCTACCTGCCAGCGACAACCACCGGGTGGAGGGCACAGCATGGTCCCAGGCAAC  
CCTGGTGGCCGGGGCACGGGGCGGGAGGCATGGGCCGGCAGGGACCTCGGTGGAGCTT  
GGTTTACAACAGGGAATAGTTTACACAAAGCATTGTCCACAGGGCTGGACTACTCCCCG  
CCCAGCGTCCCAGGTCGTCCCCCGTGGCCACCACGCTGCCTGCCGCGTACGCCACCCCG  
CAGCCAGGGACCCGGTGTCCCCGTGCAGTACGCTCACCTGCCGCACACCTTCCAGTTC  
ATTGGGTCTCCAATACAGTGGAACTATGCCAGCTTCAATCCATCACAGCTGATCCCC  
CCAACCGCAACCCCGTACCAGTGCAGTGGCCTCGGCCGAGGGGCCACCACTCCATCC  
CAGCGCTCCCAGCTGGAGGCTATTCCAATCTGCTGGCCAACATGGGCAGTCTGAGCCAG  
ACGCCGGGACACAAGGCTGAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGC  
CAGCATCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGC  
GCTCCGGGGTCAACCCCCGGGTCCCCCCCACCAGCCAGCAGAACCAGTACGTCCAC  
ATTTCCAGTTCTCCGAGAACACCGGCCGACCGCCTCTCTCCGGCCATCCCCGTCCAC  
CTCCACCCACCAGACGATGATCCCACACACGCTCACCTGGGGCCCCCTCCCAGGTC  
GTCATGCAATACGCCGACTCCGGCAGCCACTTTGTCCCTCGGGAGGCCACCAAGAAAGCT  
GAGAGCAGCCGGTGCAGCAGGCCATCCAGGCCAAGGAGGTCTGAACGGTGAGATGGAG  
AAGAGCCGGCGGTACGGGGCCCCGTCTCAGCCGACCTGGGCCTGGGCAAGGCAGGCGGC  
AAGTCGGTTCTCACCCGTACGAGTCCAGGCACGTGGTGGTCCACCCGAGCCCCTCAGAC  
TACAGCAGTCGTATCCTTCGGGGTCCGGGCTCTGTGATGGTCTGCCAACAGCAAC  
ACGCCCCGAGTACCTGGAGGTGCAACAGGCCACTCATCGTGAAGCCTCCCCTTCTACC  
CTCAACGACAAAAGTGGCCTGCATTTAGGGAAGCCTGGCCACCGGTCTACGCGTCTCA  
CCCCACACGGTCAATTCAGACCACACAGTGTCTCAGAGCCACTCCCGGTGGGACTGCCA  
GCCACGGCCTTACGCAGGACTCAACCCCTGTATCGGCTACCTGAGCGGCCAGCAG  
CAAGCAATCACCTACGCCGCGAGCTGCCCCAGCACCTGGTGATCCCCGGCACACAGCCC  
CTGCTCATCCCGGTCCGCGAGCACTGACATGGAAGCGTCCGGGGCAGCCCCGGCCATAGTC  
ACGTATCCCCCAGTTTGTGTCAGTGCCTCACACGTTGTCACCAACCGCCCTTCCCAAG  
AGCGAGAACTTCAACCCTGAGGCCCTGGTCAACCCAGGCCCTACCCAGCCATGGTGCAG  
GCCAGATCCACCTGCCTGTGGTGCAGTCCGTGGCCTCCCCGGCGGGCTCCCCCTACG  
CTGCCTCCCTACTTCATGAAAGGCTCCATCATCCAGTTGGCCAACGGGGAGCTAAAGAAG  
GTGGAAGACTTAAAAACAGAAGATTTATCCAGAGTGCAGAGATAAGCAACGACCTGAAG  
ATCGACTCCAGCACCGTAGAGAGGATTGAAGACAGCCATAGCCCCGGGCGTGGCCGTGATA  
CAGTTCGCCGTCCGGGAGCACCGAGCCAGGTCAGCGTTGAAGTTTTGGTAGAGTATCCT  
TTTTTTGTGTTTGGACAGGGCTGGTCACTCTGCTGTCGGAGAGAACCAGCCAGCTTTT  
GATTTGCCGTGTTCCAACTCTCAGTTGGGGATGTCTGCATCTCGCTTACCCTCAAGAAC  
CTGAAGAACGGCTCTGTTAAAAAGGGCCAGCCCGTGGATCCCGCCAGCGTCTGCTGAAG  
CACTCAAAGGCCGACGGCCTGGCGGGCAGCAGACAGGTATGCCGAGCAGGAAAACGGA  
ATCAACCAGGGGAGTGCCAGATGCTCTCTGAGAATGGCGAACTGAAGTTTCCAGAGAAA  
ATGGGATTGCCTGCAGCGCCCTTCTCACCAAAATAGAACCAGCAAGCCCGCGGCAACG  
AGGAAGAGGAGGTGGTCCGCGCCAGAGACCGCAACTGGAGAAGTCAGAAGACGAACCA  
CCTTTGACTCTTCTAAGCCTTCTAATTCTCAGGAGGTTAAGATTTGCATTGAAGGC  
CGGTCTAATGTAGGCAAGTAGAGGCAGCGTGGGGGAAAGGAAACGTG

**Restriction Sites:** Please inquire  
**ACCN:** NM\_000332  
**Insert Size:** 2500 bp

<b>OTI Disclaimer:</b>	Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).
<b>OTI Annotation:</b>	The ORF of this clone has been fully sequenced and found to be a perfect match to NM_000332.2.
<b>Components:</b>	The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).
<b>Reconstitution Method:</b>	<ol style="list-style-type: none"> <li>1. Centrifuge at 5,000xg for 5min.</li> <li>2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.</li> <li>3. Close the tube and incubate for 10 minutes at room temperature.</li> <li>4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.</li> <li>5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.</li> </ol>
<b>RefSeq:</b>	<u>NM_000332.2, NP_000323.2</u>
<b>RefSeq Size:</b>	10692 bp
<b>RefSeq ORF:</b>	2448 bp
<b>Locus ID:</b>	6310
<b>UniProt ID:</b>	<u>P54253</u>
<b>Cytogenetics:</b>	6p22.3
<b>Domains:</b>	AXH

**Gene Summary:**

The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegenerative disorders characterized by progressive degeneration of the cerebellum, brain stem and spinal cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. ADCA I is genetically heterogeneous, with five genetic loci, designated spinocerebellar ataxia (SCA) 1, 2, 3, 4 and 6, being assigned to five different chromosomes. ADCA II, which always presents with retinal degeneration (SCA7), and ADCA III often referred to as the 'pure' cerebellar syndrome (SCA5), are most likely homogeneous disorders. Several SCA genes have been cloned and shown to contain CAG repeats in their coding regions. ADCA is caused by the expansion of the CAG repeats, producing an elongated polyglutamine tract in the corresponding protein. The expanded repeats are variable in size and unstable, usually increasing in size when transmitted to successive generations. The function of the ataxins is not known. This locus has been mapped to chromosome 6, and it has been determined that the diseased allele contains 40-83 CAG repeats, compared to 6-39 in the normal allele, and is associated with spinocerebellar ataxia type 1 (SCA1). Alternative splicing results in multiple transcript variants, with one variant encoding multiple distinct proteins, ATXN1 and Alt-ATXN1, due to the use of overlapping alternate reading frames. [provided by RefSeq, Nov 2017]

Transcript Variant: This variant (1) encodes multiple distinct proteins due to the use of alternative translation initiation codons. The longer 815 aa protein (ATXN1, PMID:23705062) represents the canonical protein, while a shorter protein (Alt-ATXN1, PMID:23705062) that uses a different reading frame has also been described. This RefSeq represents the longer protein, ATXN1. Variant 2 also encodes ATXN1. Sequence Note: This RefSeq record was created from transcript and genomic sequence data because no single transcript was available for the full length of the gene. The extent of this transcript is supported by transcript alignments.